

## NCBI Map Viewer Tutorial

NCBI's Map Viewer can be used to visualize an organism's genome and annotation. The organisms represented in the Map Viewer include human, mouse, rat, zebrafish, mosquito, nematode, fruit fly, yeast, arabidopsis and others.

In these exercises, we will use the human genome Map Viewer to

- Locate a human gene
- Download the gene sequence along with its upstream sequence (to analyze promoter regions)
- Determine whether the sequence is in the draft or finished form
- Find a possible splice variant using Model Maker
- Identify whether variations in the gene are associated with a disease
- Obtain information about the mouse and rat homologs
- Obtain phenotype information

The first exercise includes screenshots of the suggested steps as well as a few introductory slides.

### Exercise 1

We will use as an example the glial cell derived neurotrophic factor GDNF. GDNF may contribute to Hirschsprung disease when mutated. Search for "GDNF" in the Map Viewer. Access the Human Map Viewer page by clicking on the Map Viewer link in the footer of the NCBI home page, then selecting the "*Homo sapiens* (human) Build 37.1" link. Enter GDNF as a search term and click on the "Find" button. Filter your search result for gene maps by checking the "Gene" box in the "Quick Filter" menu.

Name the chromosome on which this gene is located. Click on the Genes\_seq map link of the map element GDNF in the reference assembly. What is the map that is displayed? Turn on the ruler line for the map through the Maps & Options link. What are the nucleotide locations for the gene on the chromosome? What is the orientation of the gene on the chromosome? Download the gene sequence by using the "dl" link. (Change the strand, if necessary). In order to search for promoter elements, you may also download 5000 nucleotides of upstream sequence by adjusting the locations for the upstream 5000 nucleotides.

Add the Clone, Component and Contig maps for this region. Name the contig and GenBank accession numbers for the sequence covering this region. Are the sequences in the finished form? Is there a clone mapped to this region? If so, how can you order it?

Remove all the maps except Genes\_seq and add the Ab initio (model) and RefSeq Transcripts maps. Does the gene prediction match the current gene annotation? How many alternatively spliced transcripts have been annotated for the gene? Display the current data as "Data As Table View".

Using the Model Maker (mm), obtain a possible alternatively spliced product and its translated amino acid sequence. Search for similar proteins by using BLAST.

Remove all the maps except Genes\_seq and add the Gene maps for mouse, chimp and rat. Are the gene structures in the three organisms similar?

Remove all the maps except the human Gene map, and add the phenotype map. Name the disease with which the GDNF gene is associated. Obtain more information about the disease by linking to the corresponding OMIM record.



NCBI Map Viewer

## What is Map Viewer?

Map Viewer allows you to view and search an organism's complete genome, display chromosome maps, and zoom into progressively greater levels of detail, down to the sequence data for a region of interest.

It allows for the analysis of genes and genome annotation in graphical format.

NCBI Home GenBank BLAST

Map Viewer Home Help

The Map Viewer provides a wide variety of genome mapping and sequencing data. [More...](#)

**Search**

Search:  for:

**Tools Legend**

- Search or Browse the Genome
- BLAST
- Clone Finder
- Go to region on a chromosome
- Genome Resources page

**News**

**Human build 37 released** Aug 3, 2009  
An update to the human genome assembly and annotation is now... [more](#)

**Annotation update released for human genome build 36** Mar 24, 2008  
An annotation update for the human genome (NCBI Build 36.3) ... [more](#) [Show all](#)

**Related Resources**

- NCBI Home
- NCBI Web Search
- NCBI Site map

**Vertebrates** (17)

**Mammals** (14)

**Primates** (3)

Scientific name	Common name	Build	Tools
<i>Homo sapiens</i>	human	Build 37.1 Build 36.3	
<i>Macaca mulatta</i>	rhesus macaque	Build 1.1	
<i>Pan troglodytes</i>	chimpanzee	Build 2.1	

**Rodents** (2)

Scientific name	Common name	Build	Tools
<i>Mus musculus</i>	laboratory mouse	Build 37.1 Build 36.1	
<i>Rattus norvegicus</i>	rat	RGSC v3.4	

**Monotremes** (1)

**Marsupials** (1)

**Other Mammals** (7)

**Other Vertebrates** (3)

**Invertebrates** (12)

**Protozoa** (18)

**Plants** (46)

**Fungi** (17)

Scientific name	Common name	Build	Tools
<i>Aspergillus clavatus</i>		Build 1.1	
<i>Aspergillus fumigatus</i>		Build 2.1	
<i>Aspergillus niger</i>		Build 1.1	
<i>Candida glabrata</i>		Build 1.1	

- Locate a human gene
- Download the gene sequence along with its upstream region (for promoter analysis)
- Determine whether the sequence is in draft or finished form
- Find a possible [splice variant](#) via Model Maker
- Identify whether variations in the gene are associated with a disease
- Obtain information about mouse and rat homologues

## High Throughput Genomic Sequences

Sequencing of BACs:

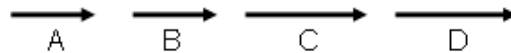
Draft:

Phase 0 – single/few pass reads of a single clone

Phase 1 – unfinished, unordered BACs with gaps



Phase 2 – unfinished, ordered with gaps

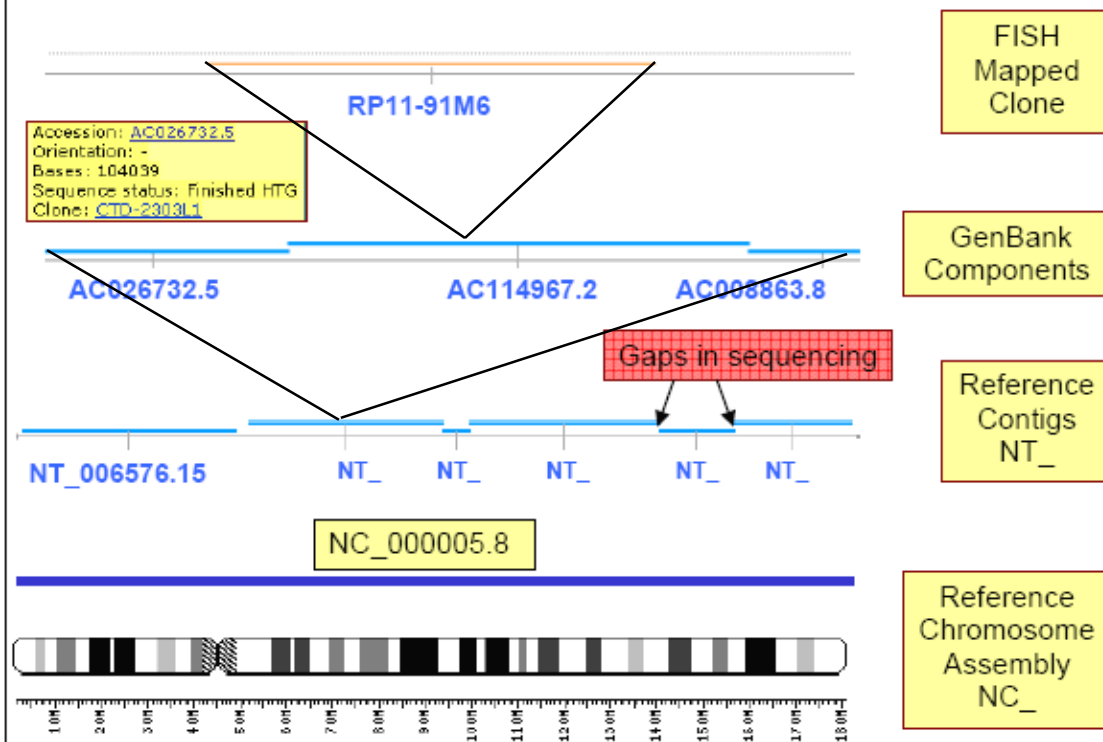


Finished:

Phase 3 – high quality finished sequence with no gaps



## Genome Assembly Accession Numbers



You are here: NCBI Write to the Help Desk

<b>GETTING STARTED</b> <a href="#">NCBI Help Manual</a> <a href="#">NCBI Handbook</a> <a href="#">Training &amp; Tutorials</a>	<b>RESOURCES</b> <a href="#">Literature</a> <a href="#">DNA &amp; RNA</a> <a href="#">Proteins</a> <a href="#">Sequence Analysis</a> <a href="#">Genes &amp; Expression</a> <a href="#">Genomes &amp; Maps</a> <a href="#">Domains &amp; Structures</a> <a href="#">Genetics &amp; Medicine</a> <a href="#">Taxonomy</a> <a href="#">Data &amp; Software</a> <a href="#">Training &amp; Tutorials</a> <a href="#">Homology</a> <a href="#">Small Molecules</a> <a href="#">Variation</a>	<b>POPULAR</b> <a href="#">PubMed</a> <a href="#">PubMed Central</a> <a href="#">Bookshelf</a> <a href="#">BLAST</a> <a href="#">Gene</a> <a href="#">Nucleotide</a> <a href="#">Protein</a> <a href="#">GEO</a> <a href="#">Conserved Domains</a> <a href="#">Structure</a> <a href="#">PubChem</a>	<b>FEATURED</b> <a href="#">GenBank</a> <a href="#">Reference Sequences</a> <a href="#">Map Viewer</a> <a href="#">Genome Projects</a> <a href="#">Human Genome</a> <a href="#">Mouse Genome</a> <a href="#">Influenza Virus</a> <a href="#">Primer-BLAST</a> <a href="#">Sequence Read Archive</a>	<b>NCBI INFORMATION</b> <a href="#">About NCBI</a> <a href="#">Research at NCBI</a> <a href="#">NCBI Newsletter</a> <a href="#">NCBI FTP Site</a>
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NCBI Home GenBank BLAST

Map Viewer Home Help

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Search

Search:

for:

Go

**Tools Legend**

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[Show all](#)

**Related Resources**

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- [NCBI Web Search](#)
- [NCBI Site map](#)

Scientific name	Common name	Build	Tools
<b>Vertebrates</b> (17)			
<b>Mammals</b> (14)			
<b>Primates</b> (3)			
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<i>Macaca mulatta</i>	rhesus macaque	Build 36.3	
<i>Pan troglodytes</i>	chimpanzee	Build 1.1	
		Build 2.1	
<b>Rodents</b> (2)			
<i>Mus musculus</i>	laboratory mouse	Build 37.1	
		Build 36.1	
<i>Rattus norvegicus</i>	rat	RGSC v3.4	
<b>Monotremes</b> (1)			
<b>Marsupials</b> (1)			
<b>Other Mammals</b> (7)			
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<i>Aspergillus niger</i>		Build 1.1	
<i>Candida glabrata</i>		Build 1.1	

NCBI NCBI Map Viewer

PubMed Nucleotide Protein Genome Gene Structure Pop

Search for  on chromosome(s)  assembly  Find

**Map Viewer**  
[Map Viewer Home](#)  
[Map Viewer Help](#)  
[Human Maps Help](#)  
[Release Notes](#)

**NCBI Resources**  
[Genome Project](#)  
[TaxPlot](#)  
[Consensus CoDing Sequence \(CCDS\)](#)  
[GRC](#)  
[Human Genome Resources](#)  
[NCBI Handbook](#)  
[RefSeq](#)  
[Trace Archive \(Watson\)](#)  
[Trace Archive \(Venter\)](#)  
[Trace FTP \(Personal Genomics\)](#)

**Homo sapiens (human) genome view**  
[Build 37.1 statistics](#) [Switch to previous build](#)

**Lineage:** [Eukaryota](#); [Metazoa](#); [Chordata](#); [Craniata](#); [Vertebrata](#); [Euteleostomi](#); [Mammalia](#); [Eutheria](#); [Euarchontoglires](#)  
[Homo](#); [Homo sapiens](#)

**August 2009:** NCBI released an updated version of the human genome reference genome assembly and updated annotation. The update includes modifications to all chromosomes and adds 9 alternate loci to the reference assembly definition provided by the [Genome Reference Consortium \(GRC\)](#). The previous version of the reference genome assembly, [NCBI Build 36.3](#), is still available for BLAST. For additional information about changes, statistics, and the status of the CCDS project please refer to: [NCBI News](#)

NCBI NCBI Map Viewer

Search for **GDNF** on chromosome(s) assembly reference Find

**Homo sapiens (human) genome view**  
Build 37.1 statistics Switch to previous build

BLAST search the human genome

Hits: 1 2 3 4 5 6 7 8 9 10 11 12 13  
41 6 12 2 16

Hits: 14 15 16 17 18 19 20 21 22 X Y HT not placed  
12 3

Search results for query "GDNF": 90 hits (assembly reference)

Chr	Match	Map element	Type	Maps
5	<a href="#">all matches</a>			
	<b>GDNF</b> family receptor alpha 3	Hs_58042	Hs_EST_Cl	Hs_UniG
	ENST00000452487	ENST00000452487	TRANSCRIPT	ensRNA
	ENST00000427982	ENST00000427982	TRANSCRIPT	ensRNA
	ENST00000381826	ENST00000381826	TRANSCRIPT	ensRNA
	ENST00000344622	ENST00000344622	TRANSCRIPT	ensRNA
	ENST00000326524	ENST00000326524	TRANSCRIPT	ensRNA
	Rattus norvegicus <b>GDNF</b> family receptor alpha 3 (Gfra3...	NM_053398.1	TRANSCRIPT	Rn_RNA
	Rattus norvegicus glial cell... <b>Gdnf</b> , mRNA	NM_019139.1	TRANSCRIPT	Rn_RNA
	Rattus norvegicus <b>GDNF</b> family receptor alpha 3, mRNA...	BC079378.1	TRANSCRIPT	Rn_RNA
	Rattus norvegicus neurotrophic... <b>GDNF</b> (Gdnf) mRNA, partial...	AF497634.1	TRANSCRIPT	Rn_RNA
	Rattus norvegicus brain <b>GDNF</b> splice variant 2 mRNA, partial...	AF205714.1	TRANSCRIPT	Rn_RNA
	Rattus norvegicus brain <b>GDNF</b> splice variant 1 mRNA, partial...	AF205713.1	TRANSCRIPT	Rn_RNA

Quick Filter  
☒ Gene  
☐ Transcript :  
☐ all  
☐ RefSeq  
☐ STS  
☐ Unigene  
 Filter

NCBI NCBI Map Viewer

Search for **GDNF AND gene[obj\_type]** on chromosome(s) assembly All Find


**Homo sapiens (human) genome view**  
Build 37.1 statistics Switch to previous build

Hits: 1 2 3 4 5 6 7 8 9 10 11 12 13  
7 3 4 3

Hits: 14 15 16 17 18 19 20 21 22 X Y HT not placed  
3 1

Search results for query "GDNF AND gene[obj\_type]": 21 hits

Chr	Assembly	Match	Map element	Type	Maps
5	reference	<a href="#">all matches</a>			
		<b>GDNF</b> family receptor alpha 3	GFRA3	GENE	<a href="#">Genes_cyto</a>   <a href="#">Genes_seq</a>
		<b>GDNF</b> : glial cell derived neurotrophic factor	GDNF	GENE	<a href="#">Genes_cyto</a>   <a href="#">Genes_seq</a>
		<b>GDNF</b> : ENSG00000168621	GDNF	GENE	<a href="#">ensGenes</a>
5	Celera	<a href="#">all matches</a>			
		<b>GDNF</b> family receptor alpha 3	GFRA3	GENE	<a href="#">craGenes</a>   <a href="#">Genes_seq</a>
		<b>GDNF</b> : glial cell derived neurotrophic factor	GDNF	GENE	<a href="#">craGenes</a>   <a href="#">Genes_seq</a>
5	HuRef	<a href="#">all matches</a>			
		<b>GDNF</b> family receptor alpha 3	GFRA3	GENE	<a href="#">Genes_seq</a>
		<b>GDNF</b> : glial cell derived neurotrophic factor	GDNF	GENE	<a href="#">Genes_seq</a>
6	reference	<b>GDNF</b> family receptor alpha like	GFRAL	GENE	<a href="#">Genes_cyto</a>   <a href="#">Genes_seq</a>
6	Celera	<b>GDNF</b> family receptor alpha like	GFRAL	GENE	<a href="#">Genes_seq</a>
6	HuRef	<b>GDNF</b> family receptor alpha like	GFRAL	GENE	<a href="#">Genes_seq</a>

NCBI  **NCBI Map Viewer**

PubMed Entrez BLAST OMIM Taxonomy

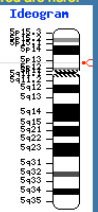
Search  Find Find in This View Advanced Search

Human genome overview page (Build 37.1)  
Human genome overview page (Build 38.3)

**Map Viewer Home**

Map Viewer Help  
Human Maps Help  
FTP  
Data As Table View  
**Maps & Options**  
Compress Map

Region Shown:  
37,798K  
37,858K

You are here:  


☐ default  
☐ master

**Homo sapiens (human) Build 37.1 (Current)**

Chromosome: 1 2 3 4 [ 5 ] 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y MT

Query: GDNF AND gene[obj\_type]

**Master Map: Genes On Sequence** [Summary of Maps](#) [Download](#)

Region Displayed: 37,798K-37,858K bp

Genes_seq	Symbol	Q	Links	E	Cyto	Description
LOC100287032	+	sv	dl ev mm	mRNA	5	similar to hCG2006445
GDNF	+	OMIM HGNC sv pr dl ev mm hm sts	CCDS SNP	best RefSeq 5p13.1-p12 glial cell derived neurotrophic factor		

Map Viewer

[http://www.ncbi.nlm.nih.gov/projects/mapview/maps\\_options.cgi?TAXID=9606&CHR=5&MAPS=gene](http://www.ncbi.nlm.nih.gov/projects/mapview/maps_options.cgi?TAXID=9606&CHR=5&MAPS=gene)

Organism: **Homo sapiens** [Help](#)

Chromosome: 5 Region Shown: 37797731.25 37857803.75

**Available Maps:**

Org: human Assembly: reference

---Sequence Maps---

- Ab initio
- Assembly
- Celera Genes
- Celera Transcripts
- Clone
- Component
- Contig
- CpG Island

**Maps Displayed (left to right):**

- [ ] Gene

([R] before map means 'ruler set')

**More Options:**


☐ Show Connections ☒ Verbose Mode

Compress Map: off

Page Length: 30

Thumbnail View: ☒ default (ideogram) ☐ master



NCBI  NCBI Map Viewer

PubMed Entrez BLAST OMIM Taxonomy

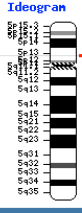
Search  Find Find in This View Advanced Search

Human genome overview page (Build 37.1)  
Human genome overview page (Build 36.3)

[Map Viewer Home](#)

Map Viewer Help  
Human Maps Help  
FTP  
Data As Table View  
[Maps & Options](#)  
[Compress Map](#)

Region Shown:  
37,798K  
37,858K

You are here:  


☐ default  
☒ master

**Homo sapiens (human) Build 37.1 (Current)**

Chromosome: [1](#) [2](#) [3](#) [4](#) [[5](#)] [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#) [MT](#)

Query: GDNF AND gene[obj\_type] [\[clear\]](#)


**Master Map: Genes On Sequence** [Summary of Maps](#) [Download](#)

Region Displayed: 37,798K-37,858K bp

Genes_seq	Symbol	Links	E	Cyto	Description
37800K					
37805K					
37810K					
37815K	<a href="#">LOC100287032</a> +	<a href="#">sv</a> <a href="#">dlev</a> <a href="#">mm</a>	mRNA	5	similar to hCG2006445
37820K					
37825K					
37830K	<a href="#">GDNF</a> +	<a href="#">OMIM</a> <a href="#">HGNC</a> <a href="#">sv</a> <a href="#">pr</a> <a href="#">dlev</a> <a href="#">mm</a> <a href="#">hm</a> <a href="#">sts</a> <a href="#">CCDS</a> <a href="#">SNP</a>	best RefSeq	5p13.1-p12	glial cell derived neurotrophic factor
37835K					
37840K					

**Homo sapiens (human) (Build 37.1)**

Region to retrieve (in chromosome coordinates):

Chromosome:  Strand:  

from:  adjust by:

to:  adjust by:

Sequence Format:

---

**This chromosome region corresponds to the contig region(s):**

Contig	start	stop	strand
NT_006576.16	37805753	37829782	+ <a href="#">Display</a> <a href="#">Save to Disk</a> <a href="#">View Evidence</a> <a href="#">ModelMaker</a>

---

**Homo sapiens (human) (Build 37.1)**

Region to retrieve (in chromosome coordinates):

Chromosome:  Strand:

from:  adjust by:

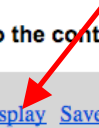
to:  adjust by:

Sequence Format:

---

**This chromosome region corresponds to the contig region(s):**

Contig	start	stop	strand
NT_006576.16	37805753	37829782	+ <a href="#">Display</a> <a href="#">Save to Disk</a> <a href="#">View Evidence</a> <a href="#">ModelMaker</a>







**Organism:** Homo sapiens [Help](#)

**Chromosome:** 5 **Region Shown:** 37797731.25 37857803.75

**Available Maps:**

Org: human Assembly: reference

Ensembl Genes  
Ensembl Transcripts  
FISH Clone (seq)  
GenBank DNA  
Gene  
Phenotype  
RefSeq Transcripts  
Repeats  
rnaBt

ADD>>  
<<REMOVE

**Maps Displayed (left to right):**

[ ] Ab initio  
[ ] RefSeq Transcripts  
[R] Gene

Change Assembly  
Move UP  
Move DOWN  
Make Master/Move to Bottom  
Toggle Ruler  
([R] before map means 'ruler set')

**More Options:**

☐ Show Connections ☒ Verbose Mode

Compress Map: off Auto Compress if > 350 px

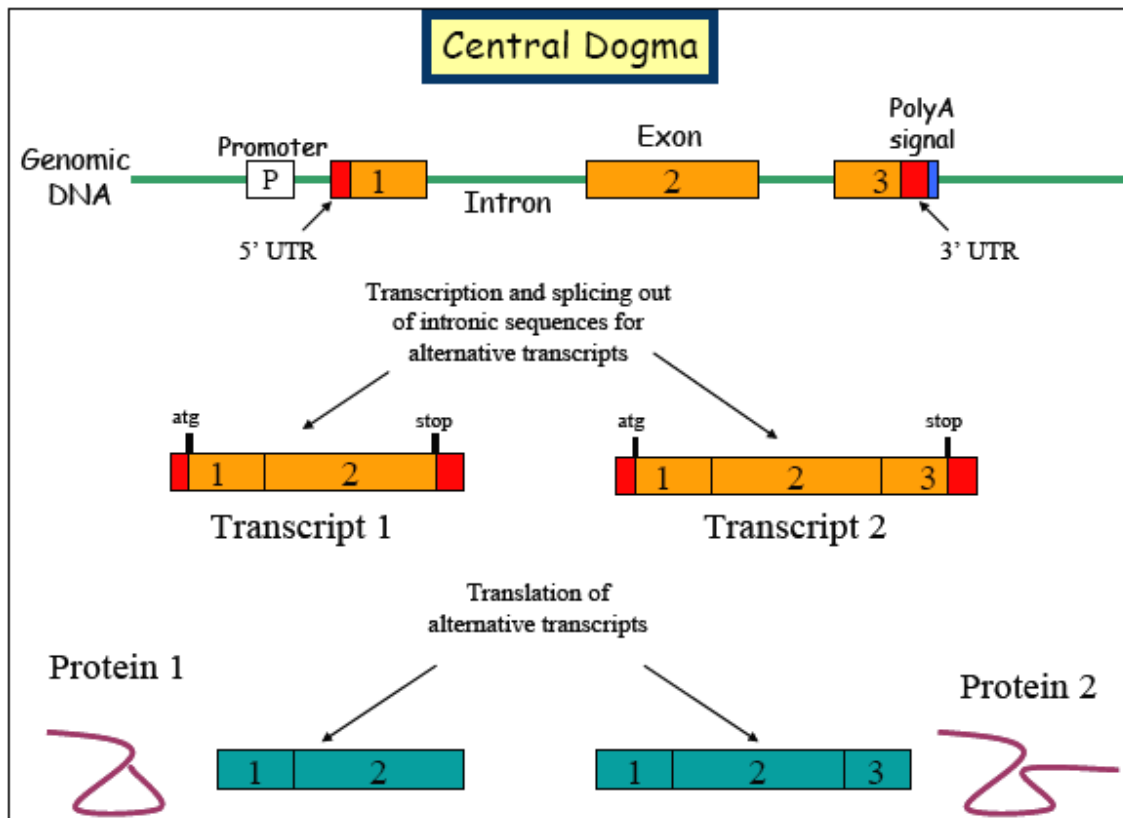
Page Length: 30

Thumbnail View: ☒ default (ideogram) ☐ master

OK Apply Close





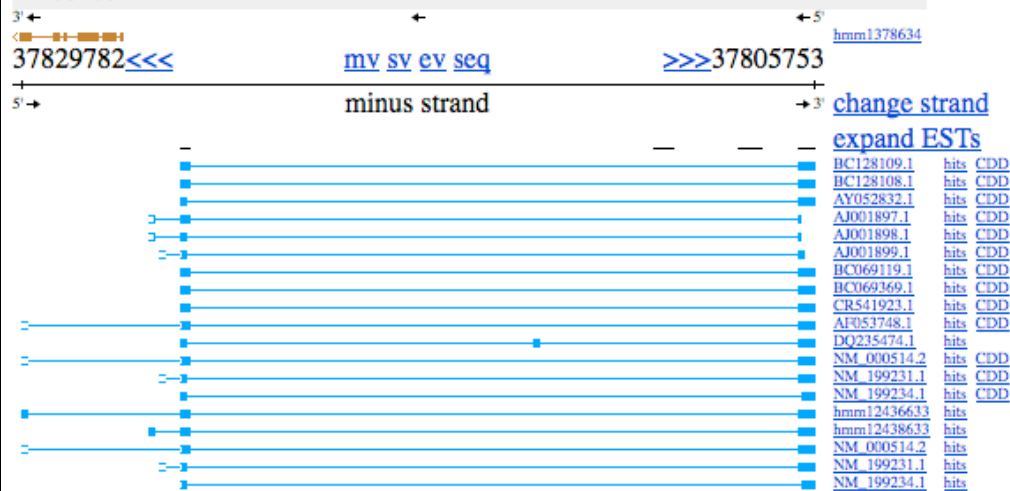


## Model Maker (Make Your Own Model by selecting an evidence [help](#) [legend](#)

exon "set" and/or add/remove individual putative exons for inclusion in your model)

Organism: Homo sapiens (Build 37.1) Chromosome: 5 Contig: [NT\\_006576.16](#) Locus: [GDNF](#)

Evidence:



Putative exons (graphic view):



Putative exons (graphic view):

Your model: 3-9-14 [clear](#)

[ORF Finder](#)  
[Save](#)

☒ Frame1, ORF= 211 [CDD](#) ☐ Frame2, ORF= 42 [CDD](#) ☐ Frame3, ORF= 22 [CDD](#)

<p>CCAAAGCGTCCGAGACTGGGTACAGTCGTCAGGCGTGACGGGGGCGGGGAGCCAGTG          ACTCCTCTGGGAGGGGAAGGGATTAGGGCCAGAATCTCTCAAAGGTGCAAAAATCCAGTC          AAGAGAGGGTTTTCGGGTATACCACGGAGGATTAAAACTTCAAGACAAGTGCCGCGCC          GGACGGGACTTTAAGATGAAGTTATGGGATGTCGTGGCTGTCTGCCTGGTGTGCTCCAC          ACCGCGTCCGCTTCCCGCTGCCCGCCGTAAGAGGCCCTCCGAGGCGCCCGCCGAAGAC</p>	<p>qsvrdwvgsrrrdggagsg*          llwegkglgpeslkgakigs          regfrvvhggklisrqpvp          dgtlr*sygMSWLSAWCCST          PRPPSRCPFVRGLPRPPKT</p>	<p>kasetgysrpgvtgargasd          ssgrgrd*ggnlsvqkssq          ervfigytted*nfgdkerrr          tgl*devMGCRCGLPGAAPH          RVRLPAARR*earqarrp</p>
---	---	---

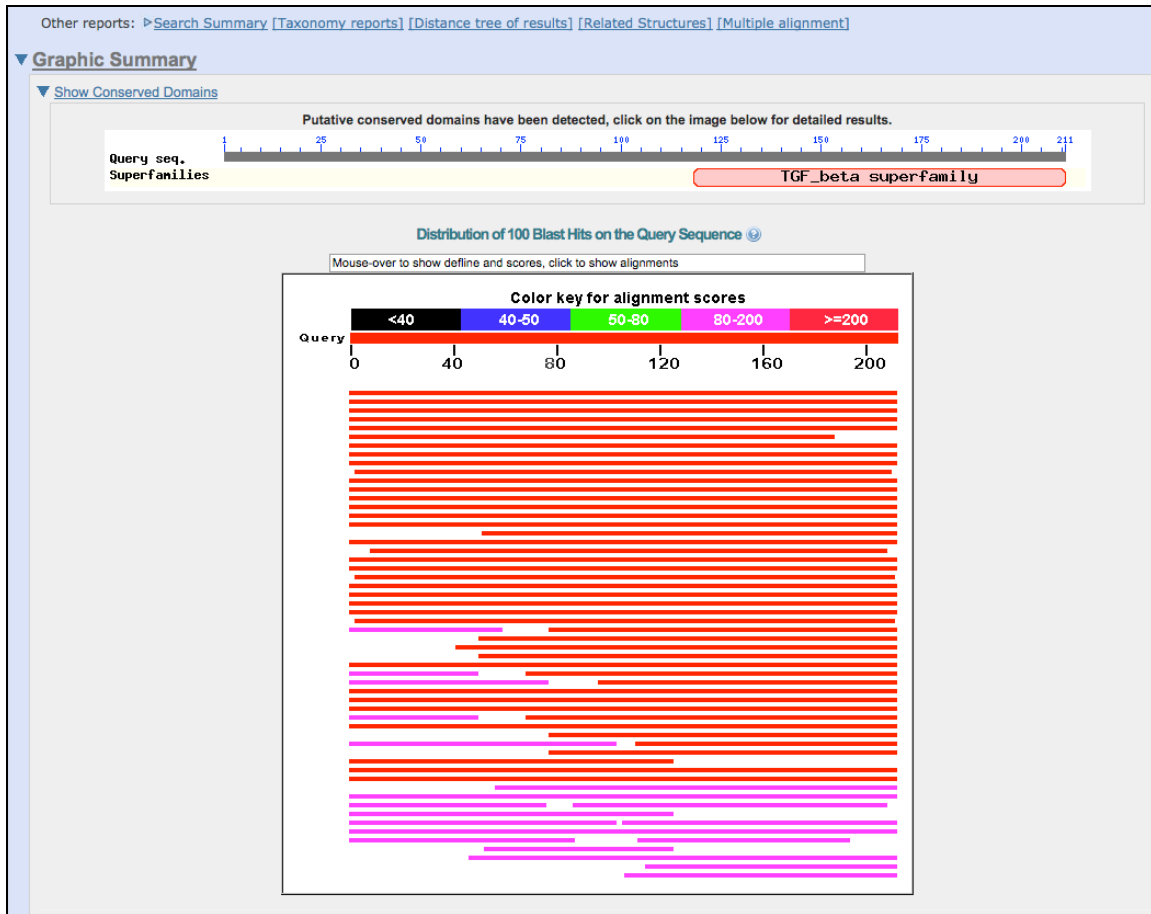
Putative exons (table view): [custom exons](#) [intron bases:](#) 2

Exon	Start	End	Sequence	ORF	Start	End	Sequence	ORF
<input type="checkbox"/> 1	CG	CCG	37829782-37829609	GAG	GT =>	9		
<input type="checkbox"/> 2	2	<= CT	CCA	37829776-37829609	GAG	GT =>	2 or 7	
<input checked="" type="checkbox"/> 3		CT	CCA	37825929-37825761	CAA	AT =>	4	
<input type="checkbox"/> 4	3	<= CT	CCA	37825929-37825736	ATG	GT =>	5 or 8 or 9	
<input type="checkbox"/> 5	4	<= AA	ATG	37825760-37825736	ATG	GT =>	9	
<input type="checkbox"/> 6		CC	CAT	37825593-37825497	TGG	GT =>	8	
<input type="checkbox"/> 7	2	<= AG	GTG	37824924-37824899	AAG	AT =>	9	
<input type="checkbox"/> 8	4 or 6	<= AG	GTG	37824924-37824826	CCG	GT =>	14	
<input checked="" type="checkbox"/> 9	1 or 4 or 5 or 7	<= AG	GTG	37824924-37824748	ACT	GT =>	12 or 14	
<input type="checkbox"/> 10		AG	ATG	37824898-37824837	CCC	GC =>	15	
<input type="checkbox"/> 11		AG	ATG	37824898-37824826	CCG	GT =>	13 or 14	
<input type="checkbox"/> 12	9	<= AG	ATG	37824898-37824748	ACT	GT =>	14	
<input type="checkbox"/> 13	11	<= AG	GGC	37814191-37814112	GAG	GT =>	14	
<input checked="" type="checkbox"/> 14	8 or 9 or 11 or 12 or 13	<= AG	CAA	37806237-37805756	ATC	TGA CT	37805753	
<input type="checkbox"/> 15	10	<= CA	GCT	37806100-37805753	TGA	CT		









### Alignments

```
>ref|XP_001143054.1| PREDICTED: similar to neurotrophic factor isoform 3 [Pan troglodytes]
gb|EAW55964.1| glial cell derived neurotrophic factor, isoform CRA_b [Homo sapiens]
Length=228
```

Score = 432 bits (1112), Expect = 4e-120  
Identities = 211/211 (100%), Positives = 211/211 (100%), Gaps = 0/211 (0%)

Query	1	MKLWDVVAVCLVLLHTASAFPLPAGKRPPEAPAEEDRSLGRRRAPFALSSDSNMPEDYPDQ	60
		MKLWDVVAVCLVLLHTASAFPLPAGKRPPEAPAEEDRSLGRRRAPFALSSDSNMPEDYPDQ	
Shjct	18	MKLWDVVAVCLVLLHTASAFPLPAGKRPPEAPAEEDRSLGRRRAPFALSSDSNMPEDYPDQ	77
Query	61	FDDVMDFIQATIKRLKRSPDKQMAVLP RRERNRQAAAA NPENSRGKGRRGQRGKNRGCVL	120
		FDDVMDFIQATIKRLKRSPDKQMAVLP RRERNRQAAAA NPENSRGKGRRGQRGKNRGCVL	
Shjct	78	FDDVMDFIQATIKRLKRSPDKQMAVLP RRERNRQAAAA NPENSRGKGRRGQRGKNRGCVL	137
Query	121	TAIHLNVTDLGLGYETKEELIFRYCSGSCDAAETTYDKILKNLSRNRLVSDKVGQACCR	180
		TAIHLNVTDLGLGYETKEELIFRYCSGSCDAAETTYDKILKNLSRNRLVSDKVGQACCR	
Shjct	138	TAIHLNVTDLGLGYETKEELIFRYCSGSCDAAETTYDKILKNLSRNRLVSDKVGQACCR	197
Query	181	PIAFDDDSLFLDDNLVYHILRKHS AKRCGCI	211
		PIAFDDDSLFLDDNLVYHILRKHS AKRCGCI	
Shjct	198	PIAFDDDSLFLDDNLVYHILRKHS AKRCGCI	228

```

>gb|AAX36770.1| glial cell derived neurotrophic factor [synthetic construct]
Length=212

Score = 432 bits (1112), Expect = 7e-120
Identities = 211/211 (100%), Positives = 211/211 (100%), Gaps = 0/211 (0%)

Query 1 MKLWDVVAVCLVLLHTASAFPLPAGKRPPPEAPAEADRLGRRRAPFALSSDSNMPEDYPDQ 60
Sbjct 1 MKLWDVVAVCLVLLHTASAFPLPAGKRPPPEAPAEADRLGRRRAPFALSSDSNMPEDYPDQ 60

Query 61 FDDVMDFIQATIKRLKRS PDKQMAVLPRRERNRQAAAANPENSRGKGRRGQRGKNRGCVL 120
Sbjct 61 FDDVMDFIQATIKRLKRS PDKQMAVLPRRERNRQAAAANPENSRGKGRRGQRGKNRGCVL 120

Query 121 TAIHLNVTDLGLGYETKEELIFRYCSGSCDAAETTYDKILKNLSRNRRLVSDKVGQACCR 180
Sbjct 121 TAIHLNVTDLGLGYETKEELIFRYCSGSCDAAETTYDKILKNLSRNRRLVSDKVGQACCR 180

Query 181 PIAFDDDLNFLVYHILRKHS AKRCGCI 211
Sbjct 181 PIAFDDDLNFLVYHILRKHS AKRCGCI 211

>ref|NP_000505.1| glial cell derived neurotrophic factor isoform 1 preproprotein
[Homo sapiens]
ref|XP_526944.1| PREDICTED: glial cell derived neurotrophic factor isoform 4 [Pan
troglodytes]

```

**Organism: Homo sapiens**
[Help](#)

**Chromosome:** 5
**Region Shown:** 37797731.25 37857803.75

**Available Maps:**

Org: rat

Ensembl Transcripts
GenBank DNA
Gene
Phenotype
RefSeq Transcripts
Repeats
rnaHs
rnaMm
rnaRn

ADD>>
<<REMOVE

**Maps Displayed (left to right):**

[R][mouse][human:5] Gene
[ ][human][5] Gene

Move UP
Move DOWN
Make Master/Move to Bottom
Toggle Ruler
([R] before map means 'ruler set')

**More Options:**

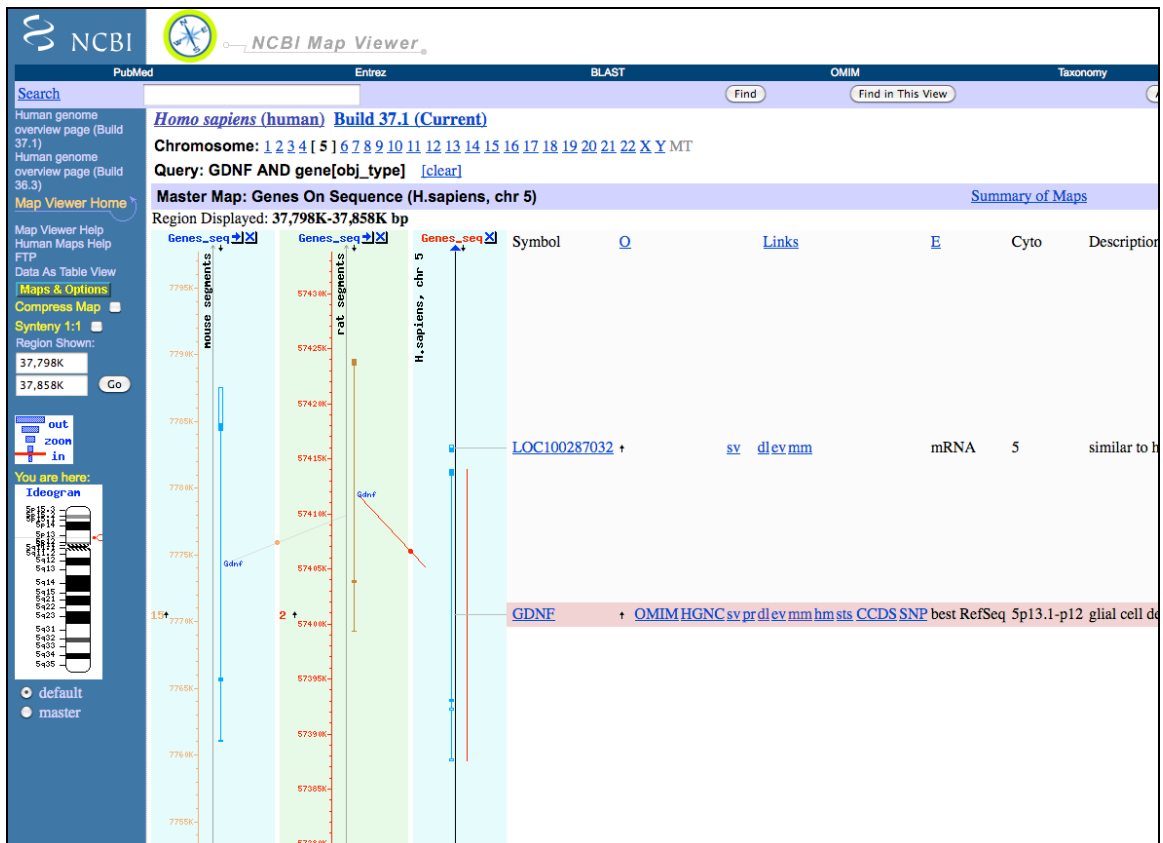
☒ Show Connections
☒ Verbose Mode

Compress Map: off
Auto Compress if > 350 px

Page Length: 30

Thumbnail View: ☒ default (ideogram) ☐ master

OK Apply Close



Organism: Homo sapiens [Help](#)

Chromosome:  Region Shown:

Available Maps: Org:  Assembly:

GenBank DNA Gene NCI Clone <b>Phenotype</b> RefSeq Transcripts Repeats STS TCAG Genes TCAG Transcripts	<input type="button" value="ADD&gt;&gt;"/> <input type="button" value="REMOVE&lt;&lt;"/>	Maps Displayed (left to right): <input type="button" value="Change Assembly"/> <input type="button" value="Move UP"/> <input type="button" value="Move DOWN"/> <input type="button" value="Make Master/Move to Bottom"/> <input type="button" value="Toggle Ruler"/> ([R] before map means 'ruler set')
--	---	---


More Options:

☐ Show Connections ☒ Verbose Mode

Compress Map:  Auto Compress if >  px

Page Length:

Thumbnail View: ☒ default (ideogram) ☐ master

NCBI  NCBI Map Viewer

PubMed Entrez BLAST OMM Taxonomy Structure

Search  Find Find in This View Advanced Search

Human genome overview page (Build 36.2) [BLAST The Human Genome](#)

Human genome overview page (Build 35.1)

Map Viewer Home

Map Viewer Help Human Maps Help FTP Data As Table View

Maps & Options

Compress Map ☐

Region Shown: 37,833K 37,894K Go

out zoom in

You are here Ideogram

default master

**Homo sapiens Build 36.2 (Current)**

Chromosome: [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#) [MT](#)

Query: GDNF AND gene[*obj\_type*] [\[clear\]](#)

Master Map: Genes On Sequence [Summary of Maps](#) [Maps & Options](#)



Region Displayed: 37,833K-37,894K bp [Download/View Sequence/Evidence](#)

Pheno [\[X\]](#) Genes\_seq [\[X\]](#) Symbol [\[O\]](#) [Links](#) [E](#) Cyto Description

37835K  
37840K  
37845K  
37850K  
37855K  
37860K  
37865K  
37870K  
37875K

Symbol: GDNF  
Description: glial cell derived neurotrophic factor

GDNF + OMM HGNC: sv pr dl ev mm hm sts CCDS SNP best RefSeq 5p13.1-p12 glial cell derived neurotrophic factor

NCBI  OMIM  Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome Structure PMC OMIM

Search OMIM  for  Go Clear

Limits Preview/Index History Clipboard Details

Display Detailed  Show 20  Send to

**\*600837** [GeneTests, Links](#)

**GLIAL CELL LINE-DERIVED NEUROTROPHIC FACTOR; GDNF**


**Alternative titles; symbols**

HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 3, INCLUDED; HSCR3, INCLUDED

Gene map locus [5p13.1-p12](#)

**TEXT**

**CLONING**

[Lin et al. \(1993\)](#) isolated a specific dopaminergic neurotrophic protein, designated 'glial cell line-derived neurotrophic factor' (GDNF), from a rat B49 glial cell line. The corresponding cDNA was cloned from both human and rat cDNA libraries. The predicted 211-amino acid sequences of the 2 proteins showed 93% homology. The human GDNF precursor is processed to a mature 134-amino acid protein with 2 potential N-linked glycosylation sites; it exists as a homodimer. The mature protein contains 7 conserved cysteine residues spaced similarly to members of the TGF-beta superfamily (see [190180](#)). 

**MAPPING**

[Schindelhauer et al. \(1995\)](#) mapped the GDNF gene to human chromosome 5p13.1-p12 by fluorescence in situ hybridization (FISH). By study of a cell hybrid panel and by FISH, [Bermingham et al. \(1995\)](#) mapped the GDNF gene to 5p13.3-p13.1.

**GENE FUNCTION**

[Lin et al. \(1993\)](#) found that recombinant human GDNF specifically promoted the survival and differentiation of dopaminergic neurons in rat embryonic midbrain cell cultures. GDNF also enhanced high-affinity uptake of dopamine in these cells. No effect of GDNF was seen on GABAergic or serotonergic neurons or astrocytes. [Schaar et al. \(1993\)](#) identified Gdnf transcripts in the substantia nigra and in type 1 astrocytes of rat basal

## Exercise 2

Search for "PRNP" in the Map Viewer. Access the Human Map Viewer page by clicking on [Map Viewer](#), then selecting the "*Homo sapiens* (human) Build 36" link. Enter PRNP as a search term.

Name the chromosome on which this gene is located. Click on the Genes\_seq map link of the map element PRNP in the reference assembly.

What is the map that is displayed? Turn on the ruler line for the map through the Maps & Options link. What are the nucleotide locations for the gene on the chromosome? What is the orientation of the gene on the chromosome?

Download the gene sequence by using the "dl" link. (Change the strand, if necessary). In order to search for promoter elements, you may also download 5000 nucleotides of upstream sequence by adjusting the locations for the upstream 5000 nucleotides.

Add the Clone, Component and Contig maps for this region. Name the contig and GenBank accession numbers for the sequence covering this region. Are the sequences in the finished form? Is there a clone mapped to this region? If so, how can you order it?

Remove all the maps except Genes\_seq and add the Ab initio (model) and RefSeq Transcripts maps. Does the gene prediction match the current gene annotation? How many alternatively spliced transcripts have been annotated for the gene? Display the current data as "Data As Table View".

Using the Model Maker (mm), obtain a possible alternatively spliced product and its translated amino acid sequence. Search for similar proteins by using BLAST.

Remove all the maps except Genes\_seq and add the Gene maps for mouse, chimp and rat. Are the gene structures in the three organisms similar?

Remove all the maps except the human Gene map, and add the phenotype map. Name the disease with which the PRNP gene is associated. Obtain more information about the disease by linking to the corresponding OMIM record.